Table of Disorders Screened by the Program

Condition	Incidence	Symptoms if not Detected	Treatment
Congenital Hypothyroidism: A condition in which the thyroid gland cannot make enough thyroid hormone for normal body and brain growth.	1 in 4,485 births (1 in 2,655 births in Hawaii)	Mental retardation, other brain damage, growth delay	Thyroid hormone replacement
Congenital Adrenal Hyperplasia (CAH): A condition in which the adrenal glands are unable to produce normal amounts of certain hormones.	1 in 13,700 births (1 in 36,285 births in Hawaii)	Salt wasting, dehydration, shock in infants Abnormal genital organs in females	Glucocorticoid and/or mineralcorticoid
Hemoglobinopathies (including Sickle Cell): Conditions in which abnormal hemoglobin in red blood cells may cause anemia	Sickle cell disease: 1 in 15, 000 births (1 in 21,771 births in Hawaii)	Sickle cell disease: Anemia, painful crises, death	Sickle cell disease: Penicillin
Biotinidase Deficiency: A condition in which the body is unable to use biotin, a B vitamin.	1 in 60,000 births (1 in 27,214 births in Hawaii)	Mental retardation, seizures, skin rash, loss of hair, death	Supplement with biotin
Galactosemia: A condition in which the body cannot break down a sugar (galactose) found in milk.	1 in 60,000 births (No cases of classic galactosemia in Hawaii)	Severe brain damage, kidney damage and eye abnormalities in neonates, death	Strict galactose-free diet

Chart of Metabolic Disorders Screened
www.state.hi.us/health/resource/family/genetics/metatable

Condition	Incidence	Symptoms if not Detected	Treatment
Homocystinuria: A condition in which the body cannot break down several amino acids in protein foods	1 in 200,000 births	Heart disease, stroke, possible mental retardation, psychiatric problems	Low methionine diet Supplement with pyridoxine, L-cysteine, and betaine
Maple Syrup Urine Disease (MSUD): A condition in which the body cannot break down several amino acids in protein foods.	1 in 150,000 births (1 in 27,214 births in Hawaii)	Neonatal coma, convulsions, mental retardation, death	Diet low in branched chain amino acids
Phenylketonuria (PKU): A condition in which the body cannot break down one of the amino acids found in protein foods	1 in 15,900 births (1 in 36,285 births in Hawaii)	Severe mental retardation, seizures	Low phenylalanine diet
Tyrosinemia Types I and II: A condition in which the body cannot break down several amino acids in protein foods	1 in 100,000 births (1 in 1,846 French Canadian births	Liver disease, kidney problems, seizures, rickets	Low phenylalanine and tyrosine diet Liver transplant if necessary
Short Chain acyl-CoA Dehydrogenase Deficiency (SCAD): A condition in which the body cannot break down dietary fats to make energy.	Rare	Developmental delay, muscle weakness Can have no symptoms or problems	Diet low in fats Supplement with carnitine
Medium Chain acyl-CoA Dehydrogenase Deficiency (MCAD): A condition in which the body cannot break down dietary fats to make energy.	1 in 15,000 births (More common in Northern Europeans)	Development delay, seizures, coma, sudden death	Avoid fasting, low fat diet Supplement with carnitine and cornstarch

Condition	Incidence	Symptoms if not Detected	Treatment
Long Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD): A condition in which the body cannot break down dietary fats to make energy.	Rare (More common in those with Finnish ancestry)	Developmental delay, muscle weakness, possible liver failure	Avoid fasting Supplement with carnitine, cornstarch, MCT, and DHA
Very Long Chain acyl-CoA Dehydrogenase Deficiency (VLCAD): A condition in which the body cannot break down dietary fats to make energy.	Rare	Heart problems, liver problems, sudden infant death	Avoid fasting, avoid certain fatty foods Supplement with cornstarch, MCT, and possibly carnitine IV glucose during illness
Multiple acyl-CoA Dehydrogenase Deficiency (MADD): A condition in which the body cannot break down dietary fats to make energy.	Rare	Vomiting, muscle weakness, hypoglycemia	Low protein and low fat diet Supplement with riboflavin and carnitine
Carnitine Uptake/Transport Defects: A condition in which the body cannot break down dietary fats to make energy	Rare	Developmental delay, muscle weakness Possible coma and death	Avoid fasting, low fat diet Supplement with carnitine
Beta-Ketothiolase Deficiency: A condition in which the body cannot break down and get rid of certain organic acids	Rare	Recurrent, severe metabolic acidosis	Sodium bicarbonate, IV fluids Possible dialysis Supplement with carnitine

Condition	Incidence	Symptoms if not Detected	Treatment
Glutaric Acidemia Type I: A condition in which the body cannot break down and get rid of certain organic acids	1 in 30,000 livebirths (More common in people of Amish ancestry)	Neurological deterioration, muscle weakness, seizures, possible dystonic cerebral palsy Some people may have no symptoms	Restrict lysine and tryptophan in diet Supplement with riboflavin and carnitine
Isobutyryl CoA Dehydrogenase Deficiency: A condition in which the body cannot break down and get rid of certain organic acids	Very rare	Heart problems	Carnitine supplementation
Isovaleric Acidemia: A condition in which the body cannot break down and get rid of certain organic acids	1 in 50,000 births	Vomiting, lack of appetite, lethargy, neuromuscular irritability, hypothermia	Protein-restrictive diet Supplement with carnitine and glycine
Malonic Aciduria: A condition in which the body cannot break down and get rid of certain organic acids	Rare	Developmental delay, vomiting, seizures, cardiomyopathy, hypoglycemia	Avoid fasting Restrict fats in diet
Methylmalonic Acidemias: A condition in which the body cannot break down and get rid of certain organic acids	1 in 50,000 to 1 in 100,000 births	Lethargy, vomiting, dehydration, respiratory distress, muscle weakness, coma, seizures, developmental delay	Low-protein diet and/or restriction of isoleucine, valine, and threonine

Condition	Incidence	Symptoms if not Detected	Treatment
Multiple Carboxylase Deficiency: A condition in which the body cannot break down and get rid of certain organic acids	1 in 87,000 births	Seizures, immune system impairment, skin rashes, hair loss, hearing loss, mental retardation	Biotin supplementation
Propionic Acidemia: A condition in which the body cannot break down dietary fats to make energy	1 in 35,000 to 1 in 75,000 births	Mental retardation, seizures, movement disorders, coma, sudden death	Avoid fasting, low protein diet Supplement with cornstarch, carnitine, and biotin Antibiotic and human growth hormone treatment
2-Methyl-3-Hydroxybutyryl CoA Dehydrogenase Deficiency: A condition in which the body cannot break down and get rid of certain organic acids	Rare	Developmental delay	In progress
2-Methylbutyryl CoA Dehydrogenase Deficiency: A condition in which the body cannot break down and get rid of certain organic acids	Rare	Lethargy, irritability, coma	Dietary restrictions
3-Hydroxy-3-Methylglutaryl (HMG) CoA Lyase Deficiency: A condition in which the body cannot break down dietary fats to make energy	Rare	Persistent vomiting, muscle weakness, lethargy, seizures, coma	Avoid fasting, low fat, low protein, high carbohydrate diet Supplement with carnitine and glucose

Condition	Incidence	Symptoms if not Detected	Treatment
3-Methylcrotonyl CoA Carboxylase Deficiency (3MCC): A condition in which the body cannot break down and get rid of certain organic acids	Rare	Muscle weakness and atrophy, seizures, dermatological changes	Dietary restrictions Supplement with carnitine and/or biotin
3-Methylglutaconyl CoA Hydratase Deficiency: A condition in which the body cannot break down and get rid of certain organic acids	Rare	Delayed motor development, short attention span, delayed development of speech	Still in development
Arginase Deficiency: A condition in which the body cannot get rid of a toxic substance called ammonia	Rare	Developmental delay, seizures, hyperactivity, ataxia	Restrict arginine and protein in diet Supplement with amino acids other than arginine Sodium benzoate therapy
Argininosuccinate Lyase Deficiency (ASA): A condition in which the body cannot get rid of a toxic substance called ammonia	1 in 70,000	Mental retardation, potential lethal coma, seizures, anorexia, vomiting, lethargy	Retrict protein in diet Supplement with arginine
Citrullinemia: A condition in which the body cannot get rid of a toxic substance called ammonia	n/a	Mental retardation, potential lethal coma, seizures, anorexia, vomiting, lethargy	Low protein diet Sodium benzoate, phenylacetate, arginine